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March 12, 1971

MAR 16 P.M.

Dr. Victor A. McKusick
Division of Medical Genetics
Department of Medicine
The Johns Hopkins University
School of Medicine
Baltimore, Maryland 21205

Dear Dr. McKusick:

I want to thank you for your letter. I am looking forward to the meeting in June.

In answer to your question regarding pyruvate kinase deficiency. We are investigating a very large kindred. The proband has a mother who is a trained social worker. She became excited about the family and traced her pedigree to one ancestor. Few cases of overt disease are present but we have been able to check over two hundred descendants, and we are going to obtain more blood. Because of the size of the sample, a bimodal distribution is evident and from correlation of genealogical information and enzyme assay we feel confident that heterozygosity can be assigned. Our initial interest was in delineating the morbidity in heterozygotes. Therefore, we have not attempted linkage studies so far. However, the family has invited me again to check further members. Because there is modest consanguinity, albeit many generations apart, the family is interested in enzyme assays for genetic counseling reasons.

We have included the case of the boy in an article which will appear in *Advances in Pediatrics* in 1971. The data we are accumulating on the family is an interesting genealogical history in its own right, and we hope to make this available also in the future. It is quite possible that this family may present a link to other kindreds in the U.S.

We would be very grateful for any advice you may have in pursuing linkage studies. Once again, thank you for your interest.

Sincerely yours,

J. van Eys, M.D.

JVE/mgh
Enc.